

## Prevalence of Congenital Colour Vision Defects Among Young Turkish Males-Results of a Survey and Review of the Literature Dischromatopsia Among Young Turkish males

*Genç Türk Erkekleri Arasında Kalıtsal Renkli Görme Bozuklukları Sıklığının Araştırılması – Bir Taramanın Sonuçları ve Literatürün Gözden Geçirilmesi  
Genç Türk Erkeklerinde Renk Körlüğü*

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### ÖZET

**Amaç:** Bu çalışma Aksaray Devlet Hastanesi Göz polikliniğinde muayene edilen genç Türk erkek olgular içerisinde kalıtsal renkli görme bozukluklarının (KRGB) sıklığını araştıran bir çalışmadır

**Gereç ve Yöntem:** Türkiye'nin 7 farklı bölgesinden gelip üniversite eğitimleri sırasında polikliniğimizde muayene edilen sağlıklı genç erkek olgularda KRGB araştırıldı. Beşyüz üç erkek olguda bu tarama için ishihara pseudoizokromatik kartları kullanıldı.

**Bulgular:** Kırmızı yeşil renk körlüğü sıklığı %5 protanlar ve %2 deutanlar olmak üzere %7 olarak bulundu

**Sonuç:** KRGB sıklığı toplumlar ve cinsiyetler arasında değişim gösterir. Renkli görme bozukluklarının Türk nüfusundaki sıklığı daha önce çeşitli çalışmalarda bildirilmiştir. Bizim çalışmamızdaki yüzdeler de bunlara paralel ancak Güney Avrupa ülkelerinde bildirilen sonuçlardan yüksek bulunmuştur.

**Anahtar Kelimeler:** Diskromatopsi, ishihara pseudoizokromatik kartları, renk körlüğü, renkli görme, Türkiye

### ABSTRACT

**Objective:** This study surveyed the prevalence of congenital colour vision defects among Turkish young males in outpatient clinics of Ophthalmology Department-Aksaray State Hospital.

**Material and Methods:** A healthy population of male adolescents coming from 7 different regions of Turkey were investigated during their university education for the presence of congenital red-green color blindness. Ishihara pseudoisochromatic plates were used for the survey among 503 male patients.

**Results:** The prevalence of red-green colour blindness was 7%(5%protans and 2%deutans).

**Conclusion:** Inherited colour vision deficiencies (CVD) vary in prevalence by population and by sex. The frequency of colour vision defects in Turkish population has been studied in some studies previously. Ratios in our study were in parallel to other studies carried out in Turkey but higher than some reports from Mediterranean Europe.

**Keywords:** Colour blindness, colour vision, dischromatopsia, Ishihara pseudoisochromatic plates, Turkey

## INTRODUCTION

**Colour vision deficiency (CVD) or dichromatopsia** is the inability or decreased ability to perceive colour differences. There are three different types of cones with three spectrally different types of pigments in human retina (1). Defective colour vision is seen when one, two or all three (achromatism) types of cones in the retina are either missing or nonfunctional. Dichromats base their colour vision on only two pigments (2). The class of defects characterized by the absence of green cones is called deuteranopia, while those defects characterized by the absence of red cones are called protanopia and those characterized by the absence of blue cones are called tritanopia. Congenital protan and deutan defects, which are collectively termed red-green color blindness, are common, affecting about 8.0–10.0% of all men. In contrast, congenital tritan defects are rare, affecting less than 1 in 10,000 people (3).

The gene that causes colour blindness is carried on the X chromosome, making the handicap more common among men than among women, who have two X chromosomes which necessitates the inheritance from both parents to transfer the disease. The English chemist John Dalton published the first scientific paper on this subject in 1798, "Extraordinary facts relating to the vision of colours" (4,5), after the realization of his own colour blindness more than 200 years ago. Because of Dalton's work, the condition was often called **daltonism**, although this term is now used for a single type of colour blindness, called deuteranopia.

The prevalence of red-green colour blindness has been found to vary between different races, tribes and ethnic groups (6). The prevalence of colour vision defects among non-Europeans is lower than European ancestry in whom it is reported to be 6.0% for males and 0.25% for females (7). Asian males have a prevalence of colour vision defects of 4.9% compared to 0.64% in females. Individuals of African, Native American or Mexican ancestry have an even lower prevalence: 3.1% in males and 0.7% in females (8).

Panel or arrangement tests are commonly used for the evaluation of colour vision defects (9). 'The pseudoisochromatic plates' test is the most popular technique for screening and diagnosis of abnormal

colour vision. It is very effective in screening, especially for red-green colour vision deficiency. Dichromats and anomalous trichromats fail in most of the plates and only slight protans and deutans are able to read some plates correctly (10).

The prevalence of congenital red-green colour blindness among young Turkish males from different regions of the country was investigated in this study with the use of Ishihara pseudoisochromatic plates.

## MATERIAL and METHODS

We carried out a survey among 503 young healthy men, with an age range of 19-32 years ( $21.1 \pm 2.01$  years) to determine the prevalence of congenital colour blindness. The survey was carried out between January-July 2010, among the university students in Aksaray, who admitted to our outpatients' clinic with their own will to attend in this population screen. The authors adhered to the Declaration of Helsinki and all state laws in our country and the patients in our study were asked to give consent form before being enrolled in the study. This study was approved by the local ethics committee of Aksaray State Hospital, Ministry of Health. These randomly selected recruits were healthy men from different regions of Turkey. Cases who were on any kind of medication, who habitually drank or smoked, or who had a history of exposure to solvents or neurotoxic chemicals were excluded from the study.

A complete ophthalmologic examination was performed, including visual acuity testing, applanation tonometry, and dilated fundus examination. The patient then noted his geographical region of origin on a form not observed by the ophthalmologist.

The refractive errors of all subjects were corrected prior to the test. Ishihara's Test for Color Blindness (Kanehara, Tokyo) was used to examine the ability of the subjects to recognize certain numbers in 17 plates. The test was given by an ophthalmologist (KT) in the same room with sufficient indirect daylight during morning hours. The plates were viewed binocularly at a distance of 50 cm. The number of correct answers was noted as the test score for that person. A person with a test score below 7 was considered colour

blind and then classified as having either protanopia or deuteranopia (7). The person was asked to read the numbers seen on the test plates 1 to 17. An assessment of reading plates 1 to 15 was done in order to determine the normality or defectiveness of colour vision. If 13 or more plates are read correctly, the colour vision is regarded as normal. If only 7 or less than 7 plates are read correctly, the color vision was regarded as red green deficient. The plates 16 and 17 are used to differentiate protan and deutan types of colour vision efficiency (11).

The data were analyzed statistically using the Chi-square test.

## RESULTS

Among seven geographic regions of Turkey; it is known that The Marmara, The Aegean and Mediterranean regions have a low ratio of consanguinity. In the Black Sea, Eastern Anatolia and Southeastern Anatolia regions the consanguinity is reported to be high (7).

The distribution of colour blindness among our subjects is given in Table 1.

None of the subjects showed any evidence of other ocular pathologies. Using the Ishihara plates, the subjects were categorized as colour normal (n = 468), protan deficient (n =25) or deutan deficient (n =10). Thirtyfive congenitally colour blind people were detected (7%). Five percent of them had protanopia and 2%percent had deuteranopia.

In our study, most of the subjects were from the Anatolian Plateau (n = 132), whereas there was only a small group from the Marmara region (n = 54). The highest percentage of colour blindness was in the

Southeastern Platfrom region (7/71) (10%) and the lowest percentage was observed among the Aegean population (2/59) (3%). Although the differences among the prevalence of colour blindness in different regions of Turkey seemed to be high, no statistically significant difference was detected (Chi-square test, p = 0.775).

## DISCUSSION

The ability to perceive the colours distinguishes humans from other species. However, inherited congenital colour vision defects (CVD), comprising a number of distinct disorders, are relatively commonly. Most colour vision defects (after ruling out other ocular pathologies) are congenital and permanent. The X-linked disorder which results in difficulties in distinguishing between colours in the red/green spectrum is most common, while more severe types of CVD are rare.

Colour vision deficiencies can be classified as acquired or inherited (12). Inherited form is classified into three types: **monochromacy, dichromacy, and anomalous trichromacy** (12). Monochromacy, also known as “total colour blindness,” is the lack of ability to distinguish colours; caused by cone defect or absence (12). Rod monochromacy (achromatopsia) is an extremely rare, nonprogressive inability to distinguish any colours as a result of absent or nonfunctioning retinal cones. It is associated with photophobia, nystagmus, and poor vision. (12). Cone monochromacy is a rare total colour blindness that is accompanied by relatively normal vision, electoretinogram, and electrooculogram (12). Dichromacy is a moderately severe colour vision defect in which one of the three basic colour mechanisms

**Table 1: The distribution of color blindness in Turkey**

Region	Ratio of CVD	Percentage
Marmara	3/54	6%
Southeastern Anatolia	7/71	10%
Central Anatolia	11/132	8%
Black Sea	5/69	7%
Mediterranean	3/61	5%
Aegean	2/58	3%
Eastern Anatolia	4/58	7%

is absent or not functioning. It is hereditary and, in the case of Protanopia or Deuteranopia, sex-linked, affecting predominantly males (12).

Protanopia is a severe type of colour vision deficiency caused by the complete absence of red retinal photoreceptors. It is a form of dichromatism in which red appears dark. It is hereditary, sex-linked, and present in 1% of males (12). Deuteranopia is a colour vision deficiency in which the green retinal photoreceptors are absent, moderately affecting red–green hue discrimination. It is likewise hereditary and sex-linked and comprise about 5% of the male population (9,13). Tritanopia is a very rare colour vision disturbance in which there is total absence of blue retinal receptors and unlike the other forms, it is not sex-linked (12). Tritan CVD should always raise the concern for an acquired colour deficiency secondary to a disease process in the retina, optic nerve, or elsewhere in the visual pathway. The gene coding for the blue receptor, or short wavelength S-cone, has been located on chromosome 7 as a missense mutation, exhibiting autosomal dominant inheritance with incomplete penetrance; thus, phenotypic expression is shared equally by men and by women (9, 14-16).

Ishihara's Test for Colour Blindness. is the most commonly used clinical colour vision tests for screening purposes. Ishihara's plates detect red and green colour vision defects. Many other colour vision tests are also available in clinical practice. Cupelli *et al* (17) reported that candidates who pass the Ishihara test have sufficient colour perception to meet ordinary needs and further tests are not necessary. This was our rationale for using the Ishihara test in this study.

The panel tests including Farnsworth Panel D-15 and Farnsworth Munsell 100-hue tests are more accurate in classifying color deficiency. Farnsworth Munsell 100-hue test is very sensitive, the spectrum is divided into 4 parts during testing and the patient is asked to discriminate between shades of similar colours. This test is however fatiguing and time-consuming. Farnsworth Panel D-15 is a quicker test and more convenient for routine practice with a single box of 15 colored tablets. But it is not very sensitive and may miss mild cases. Cases that fail Ishihara plates but pass

Panel D-15 are probably mild cases which will not have colour discrimination problems in most jobs. On the other hand, Panel D-15 test is very effective in the discrimination of congenital and acquired defects (18). Since we only aimed to investigate the prevalence of congenital CVD's in otherwise normal individuals, we preferred to use the Ishihara plates as screening test, which is quick and reliable in population screenings (19).

The Ishihara plates contain a confusion of spots of various colours hiding wavy lines or numbers, some of which are read differently by unaffected people and those with colour blindness, or cannot be read at all by the latter. It is suggested that plates with wavy lines should preferably be used between the ages of 4 and 11, and those with numbers for ages 12 and over, but the use of the lines does lead to a higher incidence of errors (20,21). The test should be carried out, if possible, in natural daylight, avoiding direct sunlight. It can therefore be concluded that the Ishihara test is good for screening, but does not discriminate accurately between complete and partial colour blindness (22). We did all the examinations in the same room with the same lighting conditions and only one ophthalmologist interpreted the results.

The incidence of red-green colour blindness has been found to vary between different races, tribes and ethnic groups.

The results of this study show a prevalence of 7% among young Turkish males, which is higher than in Spain (4.02%), Colombia (2.53%) and Italy (5.3%) but lower than in Denmark (8.7%) and the United States (8.0%) (11, 23-26). Since consanguineous marriages and related hereditary disorders are often seen in Turkey (27), this result may be a reflection of this high consanguineous marriage rate.

There are some studies that report the prevalence of colour blindness to be 5.41 % and 7.86 %, respectively, among males in Turkey (27-31) (Table 2). These studies also used Ishihara colour plates to screen the populations although their results are controversial. A consensus for the interpretation of the test results should be established among investigators. Although the performance of the subject in reading the Ishihara plates is evaluated to determine colour-blindness,

**Table 2: Reported Prevalences of CVD in Turkish Population**

Author	Case Number		Percentage of CVD		Total (%)
	Male	Female	Male	Female	
Say et al. (1996)	1900	100	5.36	0.05	5.41
Tümerdem et al. (1985)	6644	5859	5.49	0.16	5.65
Gökbel et al. (1992)	402	307	7.21	0.65	7.86
Ayhan et al. (1994)	8382	7413	5.19	1.30	6.49
Isikli et al. (1999)	3345	2606	5.50	0.80	6.30

there is not a standard criteria for this. Some investigators diagnosed colour deficiency by using transformation plates (plates 2–9) (30) whereas others used hidden digit plates (plates 18–21) to determine colour deficiency (32). Obviously, consensus among the investigators is very important, in order to avoid controversial results.

### CONCLUSION

The slight difference of the CVD prevalences in geographic regions of our country is probably a result of genetic mixing within the population. But further studies at the molecular level are needed analysing CVD prevalences with respect to females from different regions of Turkey.

Hereditary colour vision defects cannot be treated. Although several therapies have been proposed (e.g., electrical eye stimulation, iodine injections, or large doses of vitamins), there are no proven treatments or surgical procedures that improves the colour vision (33). The main conclusion is that early diagnosis, education, and awareness of this condition are the mainstay issues of the protection. In the regions where CVD's are high, prevention of consanguineous marriages may improve the quality of life by reducing the prevalence of CVD's.

### REFERENCES

- 1- Curcio CA, Sloan KR, Kalina RE, Hendrickson AE. Human photoreceptor topography. *J Comp Neurol* 1990;292(4):497–523.
- 2- Díez MA, Luque MJ, Capilla P, Gómez J, de Fez MD. Detection and assessment of colour vision anomalies and deficiencies in children. *J Ped Ophthalmol Strabismus* 2001;38(4):195–205.

- 3- Went LN, Pronk N. The genetics of tritan disturbances. *Hum Genet* 1985;69(3):255.
- 4- Cole BL. The handicap of abnormal colour vision. *Clin and Exp Optom* 2004; 87(4):258-75.
- 5- Dalton J. Extraordinary facts relating to the vision of colours: with observations. *Memoirs of The Literary and Philosophical Society of Manchester* 1798;5 (1) :28-45.
- 6- Malhotra KC, Mutalik GS, Bhanu BV, Kate SL, Fulmali PM. The incidence of colour blindness among four endogamous nomadic groups. An example of natural selection. *Heredity* 1974;322(2):145–9.
- 7- Citirik M, Acaroglu G, Batman C, Zilelioglu O. Congenital color blindness in young turkish men. *Ophthalmic Epidemiology* 2005;12(2):133–7.
- 8- Jorgensen AL, Deeb SS, Motulsky AG. Molecular genetics of Xchromosome-linked color vision among populations of African and Japanese ancestry. High frequency of a shortened red pigment gene among Afro-Americans. *Proc Natl Acad Sci USA* 1990;87(17):6512.
- 9- Alabdelmoneam M. Prevalence of congenital color vision defects in Saudi females of Arab origin. *Optometry* 2011;82(9):543-8.
- 10- Hyon YJ, Lee JH, Wee WR. Shift of colorimetric values in ishihara pseudoisochromatic plates with plate aging. *Korean J Ophthalmol* 2005;19(2):145-8.
- 11- Shah A, Hussain R, Fareed M, Afzal M. Prevalence of red-green color vision defects among Muslim males and females of Manipur, India. *Iran J Public Health* 2013;42(1):16-24.
- 12- Morgan MJ, Adam A, Mollon JD. Dichromats detect colour-camouflaged objects that are not detected by trichromats. *Proc Biol Sci* 1992; 248(1323): 291–5.
- 13- Krill AE, Beutler E. The red-light absolute threshold in heterozygote protan carriers: possible genetic implications. *Invest Ophthalmol* 1964; 3(1):107-18.

- 14- Fitzgibbon J, Appukuttan B, Gayther S, et al. Localization of the human blue cone pigment gene to chromosome band 7q31.3-32. *Hum Genet* 1994;93(1):79-80.
- 15- Pearlman JT, Owen WG, Brounley DW, Sheppard JJ. Cone dystrophy with dominant inheritance. *Am J Ophthalmol* 1974;77(3):293.
- 16- Sandberg MA, Berson EL. Blue and green cone mechanisms in retinitis pigmentosa. *Invest Ophthalmol Vis Sci* 1977;16(2):149.
- 17- Cupelli V, Zanobini A, Frosini S, et al. General criteria of working ability in subjects with colour vision defects. *G Ital Med Lav Ergon* 2001;23(1): 21-4.
- 18- Skuta GL, Cantor LB, Weiss JS (editors). American Academy of Ophthalmology Basic and Clinical Science Course Section 12, Retina and Vitreous. In : Regillo C, Holeykamp N, Johnson MW, et al. *Retinal physiology and psychophysics. Part I. Chapter 3.* San Francisco, CA, American Academy of Ophthalmology. 2010:50-51.
- 19- Honson VJ, Dain SJ. Performance of the standard pseudoisochromatic plate test. *Am J Optom Physiol Opt* 1988;65(7):561-70.
- 20- Macfarlane JA (editor). *Progress in Child Health.* In: Hill AR. *Defective colour vision in children. Vol 1.* Edinburgh: Churchill Livingstone, 1984; 51-73.
- 21- Cox BJ. Validity of a preschool colour vision test. *J School Health* 1971;41(3):163-5.
- 22- Taylor WOG. An advisory clinic for colour defective children. *Trans Ophthalmol Soc UK* 1966;86(1):591-607.
- 23- Rebato E, Calderon R. Incidence of red-green colour blindness in the Basque population. *Anthropol Anz* 1990;48(2):145-8.
- 24- Norn M. Prevalence of congenital colour blindness among Inuit in East Greenland. *Acta Ophthalmol Scand* 1997;75(2):206-9.
- 25- Mueller WH, Weiss KM. Colour blindness in Colombia. *Ann Hum Biol* 1979;6(2):137-45.
- 26- Dickinson C, Murray I, Carden D (editors). *John Dalton Colour Vision Legacy In: Grassivaro Gallo P, Romana L, Viviani F, Camperio Ciani A. Do congenital colour vision defects represent a selective advantage?* London: Taylor & Francis, 1997; 225-234.
- 27- Ayhan B, Tumerdam Y, Ozsut H. Diskromatopsi ve genetik yapı. *Epidemiyolojik araştırma. Ist Tıp Fak Mecmuası* 1994;57(1):13-6.
- 28- Say B, Gungor E, Altay C. Türk halkında renk korlugu sikligi ve bu herediter defektlerde G6PD Eksikligi arasında munasebet. *Hacettepe Cocuk Sagligi ve Hastaliklari Dergisi* 1966;9(1):96-101.
- 29- Gokbel H, Semin I, Guvel H, et al. 4-6 yas grubu cocuklarda renk korlugu sikligi. *Dokuz Eylul Univ Tıp Fak Dergisi* 1992;6(1):55-7.
- 30- Işıkli B, Kalyoncu C, Metintaş S, Aslantaş D, Unsal A. 6-17 yas okul cocuklarında kirma kusuru ve renk korlugu. *Turkiye Tıp Dergisi* 1999; 6(1):29-32.
- 31- Tümerdem Y, Alpay T, Ayhan B. Türk cocuklarında dogumsal renk korlukleri (Epidemiyolojik bir araştırma II). *Istanbul Tıp Fakültesi Mecmuası* 1985; 48 (1):611-7.
- 32- Birch J. Efficiency of the Ishihara test for identifying red-green color deficiency. *Ophthalmic and Physiological Optics* 1997; 17(1):403-8.
- 33- Richer S, Adams AJ. Development of quantitative tools for filter aided dichromats. *Am J Optom Physiol Optics* 1984;61(4):246-55.